Prevalence of Congenital Anomalies in Neonates and Associated Risk Factors in a Tertiary Care Hospital in Eastern India

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ABSTRACT

Background: Congenital anomalies are a major cause of stillbirths and neonatal mortality. The pattern and prevalence of congenital anomalies may vary over time or with geographical location. **Aims and Objectives:** The aim of this study is to determine the proportion and types of congenital anomalies in live newborns and to study maternal and perinatal risk factors. **Materials and Methods:** This cross-sectional descriptive study was carried out in the neonatal care unit of R. G. Kar Medical College and Hospital during the period of September 2011 to August 2012. All the live born babies born in this hospital during this period were included. The newborns were examined for the presence of congenital anomalies and mothers were interviewed for socio-demographic variables. **Results:** During the study period, 12,896 babies were born, of which 286 had congenital malformations, making the prevalence 2.22%. Most of the women (55.7%) belonged to the age group between 21 and 30 years. Congenital anomalies were seen more commonly (3.3%) in the multiparas in comparison with primiparas (1.8%). The predominant system involved was Musculo-skeletal system (33.2%) followed by gastro-intestinal (GI) system (15%). Talipes (17.1%) was the most common one in musculoskeletal group and likewise cleft lip and cleft palate in GI system. Congenital anomalies were more likely to be associated with low birth weight, prematurity, multiparity, consanguinity and cesarean delivery. **Conclusion:** Public awareness about preventable risk factors is to be created and early prenatal diagnosis and management of common anomalies is strongly recommended.

Key words:
Congenital anomaly, prematurity, prevalence, risk factors, types

INTRODUCTION

According to the World Health Organization (WHO) document of 1972, the term congenital malformations should be confined to structural defects at birth.[1] However, as per the more recent WHO fact-sheet of October 2012, congenital anomalies can be defined as structural or functional anomalies, including metabolic disorders, which are present at the time of birth.[2] Congenital anomalies are an important cause of neonatal mortality both in developed and developing countries. It accounts for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.[3,4] It is not only a leading cause of fetal loss, but also contributes significantly to preterm birth, childhood and adult morbidity along with considerable repercussion on the mothers and their families.

AIMS AND OBJECTIVES

This study was undertaken to determine the proportion and pattern of congenital anomalies in live newborns and to study the associated maternal and perinatal risk factors.

MATERIALS AND METHODS

This cross-sectional descriptive study was carried out in the neonatal care unit of a tertiary care Hospital in Kolkata during the period of August 2011 to July 2012. All the babies born with congenital anomalies during this period were included. All still borns were excluded from this study.

The newborns were examined and assessed systematically for the presence of congenital anomalies. Diagnosis of congenital anomalies was based on clinical evaluation of newborn babies by the pediatrician and other appropriate investigations such as radiography, ultrasonography, echocardiography and chromosomal analysis etc., System wise distribution of the anomalies was performed. For each case, a detailed antenatal and maternal history including the

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age of the mothers, parity or the history of consanguinity were obtained by reviewing the maternal and labour ward records and by interviewing the parents.

A marriage has been considered consanguineous, when that is found to have occurred between a male and a female who are blood-related, e.g., between brother and sister, between 1st cousins etc., Birth weights >2.5 kg were considered to be normal; whereas, birth weights <2.5 kg and <1.5 kg were termed as low birth weight (LBW) and very low birth weight (VLBW) respectively. Babies born at <37 completed weeks (i.e., <259 days), calculated from the 1st day of last menstrual period, were considered as premature.

Data was entered into excel data sheet and appropriate statistical analysis was performed. Proportion was calculated and the association was tested with Chi-square test and Fisher's exact test. $P < 0.05$ was considered to be statistically significant.

RESULTS

During the study period, 12896 newborns were born in our institution; of which 286 had congenital malformations, making the prevalence 2.22%. Among all the newborns, 56 babies were born of twin delivery, three of triplet delivery and five of these 59 babies, that were products of multiple gestations, had one or more congenital anomalies. The congenital anomalies affected significantly higher proportion of male babies (2.9%) than their female counterparts (1.5%).

The predominant system involved was Musculo-skeletal system (33.2%) followed by gastro-intestinal (GI) system (15%) and central nervous system (CNS) (11.2%) [Table 1]. Talipes (17.1%) was the most common anomaly seen in the musculoskeletal group and likewise cleft lip (6.6%) and cleft palate (3.5%) in GI system and meningomyeleceole (6.3%) in CNS.

Regarding the parity of the mothers, 9,185 were primiparas and rest 3,425 were multiparas. Cases of congenital anomaly were found in 3.3% of multiparas, whereas in primiparas, the proportion was only 1.8%. It has been seen that more than half of the mothers were aged between 20 and 30 years (55.7%) with only 10% of the mothers were over the age of 30 years. The prevalence of congenitally anomalous babies born was 1.9% for mothers <20 years, 2.4% for 20-30 years and 2.2% for >30 years. However, this difference was not statistically significant. In the present study, 5 mothers had a history of consanguinity and two of them showed some congenital anomaly (40%) in their babies, whereas in non-consanguineous couples, the prevalence was only 2.2%. This percentage was about 18 times less than in consanguineous couples and was highly significant. Prematurity and LBW was found to have a higher risk of congenital anomalies. The occurrence was about 4.5 times more in case of preterm delivery as compared with the term ones, making it statistically significant. Mode of delivery was also significantly associated with congenital anomaly and it was more in case of cesarean deliveries [Table 2].

DISCUSSION

The pattern and prevalence of congenital anomalies may vary over time or with geographical location, reflecting a complex interaction of known and unknown genetic and environmental factors including socio-cultural, racial and ethnic variables.[5] With improved control of infections and nutritional deficiency diseases, congenital malformations have become important causes of perinatal mortality in developing countries like India.[6]

In the present study, the prevalence of congenital malformations in the newborns were 2.22%, which is comparable with the earlier studies from India, which reported incidence of 2.72% and 1.9%.[7,8] There are other reports from different parts of the world representing different frequency of congenital malformations.[9,10] Although we got nearly the same result as reported in other studies,[7-11] the prevalence of congenital anomaly would have been more than the present rate, if we could have included the abortions and stillbirths. Tertiary care hospital usually do not have definite catchment area and complicated cases are more commonly encountered. Hence, prevalence calculated in this type of hospital-based study cannot be projected to the total population. Community based study should be ideal for true estimation of incidence of congenital anomalies in a population.

With regard to pattern of congenital anomalies in the study, the most common system involved was musculoskeletal system (33.2%), followed by gastro-intestinal tract (GIT) (15%), CNS (11.2%), genitourinary (10.5%), cardiovascular system (9.1%), skin (8.7%) etc., This was comparable with studies conducted by others.[12-17] Some studies however recorded higher incidence of CNS malformations followed by GIT and musculoskeletal system,[9,18] whereas Suguna Bai *et al*.[19] reported GI malformations as the most common one.

More male babies with congenital anomalies than females were noted in the present study. Male preponderance was similar to the other studies.[6,7] It may be because of the fact that the females were afflicted with more lethal congenital malformations and could not survive to be born with signs of life.

Table 1: System wise distribution of congenital anomalies (*n*=286)

System	Number	Percentage
Musculo-skeletal system	95	33.2
CTEV	49	17.1
Calcaneo-valgus	6	2.1
Polydactyly	13	4.5
Syndactyly	5	1.7
Absence of depressor anguli oris	4	1.4
Absence of pectoralis major	1	0.4
Vertebral anomalies	3	1.0
Pterygium	2	0.7
Osteogenesis imperfecta	2	0.7
Phocomelia	1	0.4
Multiple defects	9	3.1
Gastro-intestinal system	43	15.0
Cleft lip	19	6.6
Cleft palate	10	3.5
Tongue tie	3	1.0
Imperforate anus	2	0.7
TEF	2	0.7
Ranula	1	0.4
Gastroschisis	2	0.7
Omphalocele	1	0.4
Duodenal atresia	1	0.4
Malrotation of gut	1	0.4
Annular pancreas	1	0.4
Central nervous system	32	11.2
Meningomyelocele	18	6.3
Encephalocele	3	1.0
Hydrocephalus	5	1.7
Anencephaly	2	0.7
Holoprosencephaly	2	0.7
Microcephaly	2	0.7
Genitourinary system	30	10.5
Hydronephrosis	14	4.9
Ambiguous genitalia	5	1.7
Posterior urethral valve	3	1.0
Polycystic kidney	3	1.0
Hypospadius	3	1.0
Epispadius	1	0.4
Extrophy of bladder	1	0.4
Cardiovascular system	26	9.1
Acyanotic	17	5.9
Cyanotic	9	3.1
Skin	25	8.7
Hemangioma	14	4.9
Skin tag	5	1.7
Aplasia cutis	2	0.7
Blueberry muffin	1	0.4
Piebaldism	1	0.4
Others	2	0.7
Syndromes	6	2.1
Down syndrome	3	1.0
Holt-Oram syndrome	1	0.4
Prune Belly syndrome	1	0.4
TAR syndrome	1	0.4
Respiratory system	5	1.8
Diaphragmatic hernia	3	1.0
Eventration of diaphragm	1	0.4
Choanal atresia	1	0.4
Multiple system affected	24	8.4

CTEV – Congenital talipes equinovarus; TEF – Tracheo esophageal fistula; TAR – Thrombocytopenia absent radius

Table 2: Association between congenital anomalies and maternal and perinatal risk factors

Variable	Groups	Congenital anomaly					
		Yes		No		Total	χ^2 value, df, *P* value
		No.	%	No.	%	No.	
Maternal age	<20 years	83	1.9	4326	98.1	4409	3.69, df=2, *P*=0.157
	20-30 years	174	2.4	7004	97.6	7178	
	>30 years	29	2.2	1280	97.8	1309	
Parity	Primiparas	171	1.8	9185	98.2	9356	23.91, *P*=0.000*
	Multiparas	115	3.3	3425	96.7	3540	
Consanguinity	Present	2	40	3	60	5	*P*=0.000*
	Absent	284	2.2	12607	97.8	12891	
Birth weight	Very low	14	0.8	1756	99.2	1770	94.17, df=3, *P*=0.000*
	Low	206	3.8	5489	96.2	5495	
	Normal	51	1.3	3747	98.7	3798	
	High	15	0.8	1818	99.2	1833	
Mode of delivery	Vaginal	205	2.5	8042	97.5	8247	7.58, *P*=0.005*
	Caesarean	81	1.7	4568	98.3	4649	
Gestation	Term	90	1	8356		8446	149.83, *P*=0.000*
	Preterm	196	4.4	4254		4450	
Gender	Male	191	2.9	6428	97.1	6619	27.97, *P*=0.000*
	Female	95	1.5	6182	98.5	6277	

*Statistically significant

Association of LBW with increased risk of congenital malformations is very well- documented.[6] Our finding is in accordance with that. The incidence of congenital anomalies was significantly higher in preterm babies as compared with the full term babies, which is in conformity with the previous studies reported from this country.[17] Mode of delivery also showed a significant association with congenital anomalies in this study with cesarean section being more commonly associated than normal delivery.

Suguna Bai *et al*.[19] reported a higher incidence of malformation in the babies born to mothers aged over 35 years, whereas Dutta *et al*.[18] documented statistically insignificant association of increased maternal age and congenital anomalies. The relationship between maternal age and babies born with congenital malformations, in our study, revealed that a majority of malformed babies were

born of mothers aged 20-29 years; though, it was statistically insignificant.

Previous studies have reported significantly higher incidence of malformations among the multiparas.[6] Our result is consistent with this finding, which indicates a positive correlation between the birth order and the incidence of congenital anomalies.

Consanguineous marriages are reported to play a major role in the occurrence of congenital malformations.[20] In the present study also, prevalence of malformed babies was more when born out of consanguineous marriages as seen in studies from Kuwait, Arab[21,22] and also India.[17]

Despite the high risk of recurrence of congenital malformations, there are no well-accepted preventive measures in developing countries like India. It indicates that strong preventive measures for congenital anomalies in this region are needed. Increasing awareness about maternal care during pregnancy, educational programs on congenital malformations and the consequences of consanguineous marriages need to be highlighted to decrease the incidence of congenital anomalies and their comorbidities.

Limitations

As it is a tertiary care hospital or referral centre, prevalence calculated may be higher than the general population in this hospital-based study. Hence, the data cannot be projected to the general population, for which population-based studies are necessary. Secondly, we could not include the abortions and stillborns, because often the abnormalities are not obvious or visible externally. In those cases, a pathological autopsy is warranted and in most of the cases, parental consent is not available for pathological autopsy.

CONCLUSION

This study has highlighted the prevalence and types of congenital anomalies seen in our locality. Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination, when needed.

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